



## C8A gene

complement C8 alpha chain

### Normal Function

The *C8A* gene provides instructions for making one piece, the alpha subunit, of a protein complex called complement component 8. The alpha subunit is linked to another piece of the complex called the gamma subunit (produced from the *C8G* gene). These two proteins interact with the beta subunit (produced from the *C8B* gene) to form complement component 8.

Complement component 8 aids in a part of the body's immune response known as the complement system. The complement system is a group of proteins that work together to destroy foreign invaders, trigger inflammation, and remove debris from cells and tissues. Complement component 8 combines with several other complement proteins to form the membrane attack complex (MAC), which inserts itself in the outer membrane of bacterial cells. This complex creates a hole (pore) in the membrane, which kills the bacterium. This part of the immune response appears to be especially important for fighting infection by bacteria in the *Neisseria* genus.

### Health Conditions Related to Genetic Changes

#### C3 glomerulopathy

#### complement component 8 deficiency

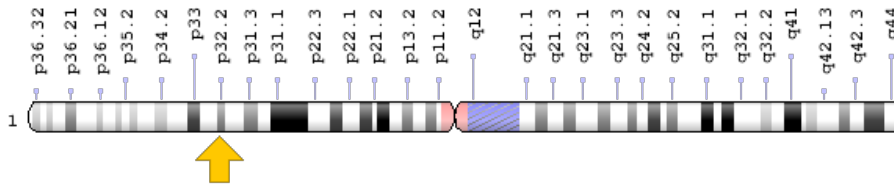
At least two mutations in the *C8A* gene have been found to cause complement component 8 deficiency type I. This condition is an immune system disorder, known as an immunodeficiency, in which the immune system is not able to protect the body effectively from foreign invaders such as bacteria. People with complement component 8 deficiency have a significantly increased risk of developing recurrent infections, particularly by *Neisseria meningitidis*, which causes meningitis, a serious condition that involves inflammation of the membranes surrounding the brain and spinal cord.

Mutations in the *C8A* gene alter the blueprint for protein production; if produced, the abnormally short alpha subunit is likely broken down quickly. The resulting shortage of this protein impairs formation of complement component 8. Deficiency of this component prevents formation of membrane attack complexes. Without this part of the immune response, affected individuals are prone to infection, particularly by *Neisseria* bacteria.

## Chromosomal Location

Cytogenetic Location: 1p32.2, which is the short (p) arm of chromosome 1 at position 32.2

Molecular Location: base pairs 56,854,770 to 56,918,221 on chromosome 1 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- complement component 8 alpha subunit
- complement component 8 subunit alpha
- complement component 8, alpha polypeptide
- complement component C8 alpha chain preproprotein

## Additional Information & Resources

### Educational Resources

- Immunobiology: The Immune System in Health and Disease (fifth edition, 2001): The Terminal Complement Proteins Polymerize to Form Pores in Membranes that can Kill Certain Pathogens  
[https://www.ncbi.nlm.nih.gov/books/NBK27100/#\\_A186\\_](https://www.ncbi.nlm.nih.gov/books/NBK27100/#_A186_)
- Molecular Biology of the Cell (fourth edition, 2002): Complement Activation Targets Pathogens for Phagocytosis or Lysis  
[https://www.ncbi.nlm.nih.gov/books/NBK26846/#\\_A4679\\_](https://www.ncbi.nlm.nih.gov/books/NBK26846/#_A4679_)

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28C8A%5BTIAB%5D%29+OR+%28complement+component+8,+alpha+polypeptide%5BTIAB%5D%29%29+OR+%28%28complement+component+8+subunit+alpha%5BTIAB%5D%29+OR+%28complement+component+C8+alpha+chain+preproprotein%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D>

### OMIM

- COMPLEMENT COMPONENT 8, ALPHA SUBUNIT  
<http://omim.org/entry/120950>

### Research Resources

- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=C8A%5Bgene%5D>
- HGNC Gene Family: Complement system  
<http://www.genenames.org/cgi-bin/genefamilies/set/492>
- HGNC Gene Symbol Report  
[http://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=1352](http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=1352)
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/731>
- UniProt  
<http://www.uniprot.org/uniprot/P07357>

### **Sources for This Summary**

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